

Newborn Screening Testing Expanded

by Mike Glass, DOH
Director, Newborn Screening Program

The State Board of Health added five new inherited disorders of metabolism to the newborn screening law (Chapter 246-650 of the Washington Administrative Code) in October 2003. Since that time, staff of the Washington Department of Health's Newborn Screening Program have worked diligently to put these new tests on-line. On January 1, 2004 biotinidase deficiency and galactosemia were added and on June 1, maple syrup urine disease, homocystinuria, and medium chain acyl-CoA dehydrogenase (MCAD) deficiency testing were added. Now, all infants born in Washington are screened for these disorders in addition to phenylketonuria (PKU), congenital hypothyroidism, congenital adrenal hyperplasia (CAH), and hemoglobinopathies (such as sickle cell disease). All of the testing is done at the Department's Public Health Laboratories in Shoreline.

Galactosemia is caused by deficiency of the enzyme galactose uridyl transferase (GALT), which is needed to convert galactose, a component of the milk sugar lactose, into glucose. GALT deficiency leads to a buildup of galactose and intermediates that are toxic. In our state, we are screening with a fluorescent assay that measures GALT activity. Screening test results are reported semi-quantitatively as Normal, Partial, or Profoundly Deficient activity.

Biotinidase deficiency, as the name describes, is a condition caused by decreased activity of the enzyme biotinidase which is needed to help the body recycle the

B12 vitamin biotin. Again, the condition is detected by measuring the level of enzyme activity, in this case with a colorimetric endpoint. As with the GALT test for galactosemia, results are reported as Normal, Partial, or Profoundly Deficient activity.

The frequency of these disorders is expected to be low, about 1 in 80,000 births for profound biotinidase deficiency and 1 in 50,000 for profound GALT deficiency leading to severe galactosemia. However, there are more prevalent, milder forms of the disorders (partial deficiency) that also benefit from treatment.

Both biotinidase deficiency and galactosemia cause severe neurological and developmental damage and sometimes death, if not detected and treated. The consequences of biotinidase deficiency are easily prevented by daily supplements of biotin, one of the B12 vitamins. Preventive treatment for galactosemia is also highly effective but more difficult because it requires strict avoidance of all milk and milk containing products.

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Practice Guidelines

The following practice guidelines have been developed by the Clinical Laboratory Advisory Council. They can be accessed at the following website:
www.doh.wa.gov/lqa.htm

Anemia	Lipid Screening
ANA	Point-of-Care Testing
Bioterrorism Event Mgmt	PSA
Bleeding Disorders	Rash Illness
Chlamydia	Red Cell Transfusion
Diabetes	Renal Disease
Group A Strep Pharyngitis	STD
Hepatitis	Thyroid
HIV	Tuberculosis
Infectious Diarrhea	Urinalysis
Intestinal Parasites	Wellness

MTS License Renewal

by Leonard Kargacin

Current Medical Test Site (MTS) licenses will expire on October 31, 2004. With over 2900 licensed facilities in Washington State, we need your timely cooperation in this process to assure that all facilities are re-licensed on time. The MTS rules require licensees to submit a completed renewal application form and fee 30 days prior to the expiration date of the license. Your application for renewal must be returned to the Office of Laboratory Quality Assurance (LQA) by **August 16, 2004** so that we can review your application, send you a fee letter, and receive your payment by October 1, 2004.

LICENSE RENEWAL PROCESS

The renewal process will be handled as follows:

1. Complete your renewal application and return it to our office **by August 16, 2004**;
2. LQA will review and process your application and send you a fee letter;
3. Upon receipt of payment, your license will be issued.

The renewal applications were mailed in Mid-July. The **pre-printed** license renewal application contains

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NOTE: Letters to the editor may be published unless specified otherwise by the author.

Website addresses:

DOH home page: <http://www.doh.wa.gov>

LQA home page: <http://www.doh.wa.gov/lqa.htm>

PHL home page:

<http://www.doh.wa.gov/EHSPHL/PHL/default.htm>

information about your medical test site from the LQA computer database.

It is essential that the information on the pre-printed renewal application be checked closely for accuracy.

The information on the renewal form is entered into the MTS computer system and the Centers for Medicare & Medicaid Services (CMS) computer system. The information in the CMS database is accessed by government reimbursement agencies, such as Medicare and Medicaid, who use the information to reimburse medical claims to providers.

TIPS FOR PROMPT RENEWAL:

- Review and **UPDATE** information on the first page. Check names, address(s), phone and fax numbers, and e-mail addresses listed. Correct as necessary.
- Add the fax number and e-mail address, if not listed.
- **SIGN** on the back of the first page.
- Indicate the tests performed as requested.
- List the volume of tests performed as requested.
- **DO NOT SEND MONEY** - we will send you a bill.
- Return completed application by **AUGUST 16, 2004**.

Questions?

- Call (206) 361-2802
- Visit our website at www.doh.wa.gov/lqa.htm
Click on the "Updates" sidebar.
Click on "2004 MTS License Renewal Instructions".
Click on "2004 MTS License Renewal Frequently Asked Questions".

NOTE: If you have not received your license renewal packet by August 2, please contact the Office of Laboratory Quality Assurance at (206) 361-2802.

**RETURN YOUR MTS
LICENSE RENEWAL
APPLICATION BY
AUGUST 16!**

Newborn Screening Testing, continued from page 1

Maple Syrup Urine Disease (MSUD) is caused by a deficiency or absence of an enzyme needed to break down the branched chain amino acids leucine, isoleucine, and valine. This results in increased serum levels of these amino acids and ketoacid intermediates. Screening is now possible using tandem mass spectrometry to measure the amino acids. If untreated, MSUD is lethal for the classical form (absence of enzyme activity) usually in the first month of life. If residual enzyme activity is present, children develop mental and physical retardation. Since therapy requires dietary restriction of branched chain amino acids, specialized medical and nutritional intervention is required. About 1 in 130,000 infants in Washington are expected to test positive for MSUD. It occurs in about 1 in 760 births to Mennonite families.

Homocystinuria is caused by the deficiency or absence of an enzyme necessary for the breakdown of the amino acid methionine that results in the build up of methionine in the blood and elevated excretion of homocystine in the urine. Screening is now possible using tandem mass spectrometry. There is a wide variation in the clinical course for affected infants. Clinical features include circulatory blood clotting (thromboembolism), and physical and mental developmental disabilities. Approximately half die by age 25 due to thromboembolism. Developmental delay and physical defects affect most. Vitamin B6 supplementation is the therapy used for those who are responsive. Dietary restriction of methionine with supplementation of cystine is used for those not responsive to Vitamin B6 therapy. With treatment, mortality and mental retardation are prevented or reduced. Prevalence of Homocystinuria varies from between 1 in 80,000 to 1 in 500,000 possibly due to the variation in sensitivity of the screening tests with age.

Medium Chain Acyl-coA Dehydrogenase Deficiency (MCADD) is caused by a defect in the production of the MCAD enzyme which functions in metabolizing fatty acids. Screening is now possible using tandem mass spectrometry to measure the build-up of intermediary compounds called acylcarnitines. There is variable expression if untreated, but fasting or infection can trigger acute episodes of hypoglycemia leading to rapid crisis or death (30% mortality following the first episode). Up to 5% of the deaths attributed to Sudden Infant Death Syndrome may, in fact, be caused by undiagnosed MCADD. Therapy includes the avoidance of fasting during illness and the reduction of dietary fat. About 1 in 10,000 infants in Washington are expected to test positive for MCADD.

Follow-up: The new screening tests have already paid off.

- On Saturday afternoon, January 31, among the several hundred tests that were processed by the Newborn Screening Program, one was positive for galactosemia. The program's follow-up section was quickly called into action. After considerable inquiry and several phone calls, the child was finally located at Mary Bridge Children's Hospital where she had been admitted for evaluation of illness of unknown origin. Follow-up staff recognized the symptoms as consistent with the severe form of galactosemia and advised that she be placed immediately on a soy-based formula. With milk removed from her diet, her acute symptoms resolved and she steadily improved throughout the day. Without screening, her prognosis would have been very uncertain; severe galactosemia is fatal for about a third of undiagnosed infants. Instead, she is now being treated and is doing well.
- A second infant tested positive for the severe form of galactosemia just a few months later. The child was again at the hospital for treatment of symptoms of "unknown origin" that were explained by the newborn screening findings.
- In May, a child was detected with the profound form of biotinidase deficiency.
- In June, just a few days after testing was implemented, an infant tested positive for MCADD. Coincidentally, this child was detected on the same day as the second child with the severe form of galactosemia.
- In addition, fourteen children have been identified with a mild form of galactosemia and seven with partially deficient biotinidase activity.

With the addition of the new disorders, Washington now screens for all nine disorders recommended by the March of Dimes.

Further information on Newborn Screening, including the new disorders, can be found on the Newborn Screening website at www.doh.wa.gov/nbs or by contacting:

Washington State Department of Health
Newborn Screening Program
1610 NE 150th Street, K17-9
Shoreline, WA 98155-0729
Phone: (206) 361-2902
FAX: (206) 361-4996
E-mail: NBS.Prog@doh.wa.gov

Eastern Washington MTS Surveyor Contact Information Update

Lori Hudson has relocated to Spokane as the Medical Test Site program surveyor for Eastern Washington. Her contact information is as follows:

Lori Hudson
Office of Laboratory Quality Assurance
1500 West 4th Avenue, Suite 305
Spokane, WA 99204-1656

Phone: (509) 458-2131
Fax: (509) 456-2997
E-mail: lori.hudson@doh.wa.gov

Calendar of Events

PHL Training Classes:

(<http://www.doh.wa.gov/EHSPHL/PHL/train.htm>)

Basic Blood Cell Morphology

September 9

Shoreline

Handling & Shipping of Biohazardous Materials

September 22

Shoreline

Northwest Medical Laboratory Symposium

October 20-23

Portland

11th Annual Clinical Laboratory Conference

November 8

Seattle

2005 WSSCLS/NWSSAMT Spring Meeting

April 28-30, 2005

Spokane

Contact information for the events listed above can be found on page 2. The Calendar of Events is a list of upcoming conferences, deadlines, and other dates of interest to the clinical laboratory community. If you have events that you would like to have included, please mail them to ELABORATIONS at the address on page 2. Information must be received at least one month before the scheduled event. The editor reserves the right to make final decisions on inclusion.

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been mailed. If you have NOT received yours by
AUGUST 2, 2004, contact (206) 361-2802.

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